

## CLAIMS:

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- A1
1. An isolated or recombinant DNA molecule encoding
    - (a) a proteoglycan expressed in retinal tissue having a membrane anchor sequence and a leucine-rich repeat motif comprising the amino acid sequence: x-x-(I, V or L)-x-x-x-x-(F, P or L)-x-x-(L or P)-x-x-L-x-x-(L or I)-x-L-x-x-N-x-(I or L) where "x" represents any amino acid, wherein an impairment in function of the proteoglycan is associated with a complete CSNB phenotype;
    - (b) the amino acid sequence of SEQ ID NO: 2;
    - (b) the amino acid sequence of SEQ ID NO: 2 with conservative amino acid substitutions; or
    - (c) an amino acid sequence which is at least 50% homologous to SEQ ID NO: 2.
  2. The DNA molecule of claim 1 which encodes an amino acid sequence which is at least 70% homologous to SEQ ID NO: 2.
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3. The DNA molecule of claim 1 wherein said DNA is cDNA.
  4. .
  5. .
  6. .
  7. .
  8. The DNA molecule of claim 1 wherein said DNA has the nucleotide sequence corresponding to SEQ ID NO:1 or naturally occurring allelic variants of SEQ ID NO:1.
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- A2
9. An isolated or recombinant polynucleotide comprising a nucleotide sequence corresponding to SEQ ID NO: 1, substantially homologous to SEQ ID NO:1 or a

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A2

nucleotide sequence that hybridizes under highly stringent conditions to a hybridization probe having a nucleotide sequence of SEQ ID NO:1 or the complement of SEQ ID NO:1.

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10. The polynucleotide of claim 9 wherein said polynucleotide is selected from the group comprising:

- (a) RNA;
  - (b) cDNA;
  - (c) genomic DNA; and
  - (d) synthetic nucleic acids.
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- A3 11. An expression vector comprising one of the DNAs or polynucleotides of claims 1, 2, 3, 8, 9 or 10.
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12. A cultured cell comprising the expression vector of claim 11.
13. A cultured cell comprising a DNA sequence or polynucleotide of one of claims 1 to 10, operably linked to an expression control sequence.
14. A cultured cell transfected with the vector of claim 11, or a progeny of said cell, wherein the cell expresses a mammalian GPI-anchored small leucine-rich proteoglycan.